

Newborn Screening Advisory Committee Meeting

Meeting Minutes of Tuesday, October 13, 2015

12:00-3:00

Wilder Center

Auditorium A - D

451 Lexington Parkway North

Saint Paul, MN 55104

Committee Members Present:

Mark Bergeron	Robert Jacobson	Dietrich Matern	Kevin Sheridan
Susan Berry	Steven Johnson	Stephen Nelson	Kathy Stagni
Vikas Bhambhani	Jan Larson	Michael Nwaneri	Jennifer Ward
Jenny Bock	Candace Lindow-Davies	Martha Overby	
Kirsten Dummer	Kristin Loncorich	Annamarie Saarinen	

Committee Members Absent: (N=notification that (s)he would be absent)

Amy Karger (N)	Amy Powers (N)	Kyriakie Sarafoglou (N)
Richard Lussky (N)	Michael Pryor	Julie Thompson Larson (N)

Guest Speakers:

Gerald Raymond, MD

Welcome and Introduction

- a. Mark McCann, Director of the Newborn Screening Program, welcomed the Committee members and guests to the Newborn Screening Advisory Committee meeting.
- b. Jan Larson, Chair of the Newborn Screening Advisory Committee, welcomed the Committee members and guests and opened the meeting with a review of the agenda.
2. Newborn Screening Updates
 - a. Maggie and Sondra referred everyone to the Newborn Screening Advisory Committee newsletter from this summer and there were no questions/discussion.
3. X-linked Adrenoleukodystrophy
 - a. Guest speaker - Gerald Raymond, MD, UMN Department of Neurology
 - b. Provided a comprehensive review of X-ALD incidence, clinical features, diagnostic work-up, and available therapies. He also provided an outlook of what newborn screening for X-ALD would look like.
4. ACHDNC and RUSP
 - a. Sue Berry (proxy for Dietrich Matern)
 - b. Background information about the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), including how it was started, background of members on the committee, how disorders are reviewed/added, what the current recommended uniform screening panel (RUSP) looks like, and what it might look like in 2016 based on recent recommendations made to the Secretary of Health and Human Services (HHS).
5. Lysosomal Disease Work Group
 - a. Sue Berry
 - b. Work group charged with providing expert guidance and recommendations to Minnesota's Advisory Committee on Heritable and Congenital Disorders on whether or

not six lysosomal disease and X-linked Adrenoleukodystrophy should be recommended for inclusion to Minnesota's newborn screening panel

- c. Work group recommends that committee aligns with the RUSP
 - i. MN NBS panel should be based on evidence
 - ii. MN should not duplicate the detailed evidence review performed by the ACHDNC
 - iii. Aligning with RUSP would result in
 - 1. Adding Pompe
 - 2. Adding Hurler (MPS-1) pending signature from HHS Secretary
 - 3. Adding X-ALD pending signature from HHS Secretary
 - d. Niemann-Pick
 - i. Not recommended for MN panel (unanimous decision); no treatment available
 - e. Fabry and Gaucher
 - i. Not recommended for MN panel (not unanimous decision)
 - 1. Early identification could improve outcomes
 - 2. Majority agree that it is outside the scope of newborn screening since symptoms arise outside of the newborn time frame
 - 3. While treatment options exist, no clear medical guidelines for how/when to treat
 - 4. Capacity to conduct effective follow-up
 - f. Krabbe
 - i. Most heavily discussed of all of the disorders under review
 - ii. Not recommended for MN Panel (not unanimous decision)
 - 1. Symptoms can develop within first 6 months but some children do not develop symptoms until later in late (juvenile, adult forms)
 - 2. Treatment is available, however not without risks – in NY 4 families elected transplant for intervention; half died as a result of transplant complications
 - 3. Despite successful transplant, children still develop symptoms- not curative
 - 4. Lack of clear medical guidelines
 - 5. Other states have indicated poor compliance in follow-up for children who are not symptomatic
6. Discussion and Vote
- a. Jan (Committee Chair) presents resolution language to be used for voting
 - i. Be it resolved that the Minnesota Department of Health Advisory Committee on Heritable and Congenital Disorders recommends to the Commissioner of Health that XXX be added to the Minnesota newborn screening panel.
 - 1. Motion was made to vote on each 8 resolutions
 - 2. Motion was seconded
 - b. Discussion
 - i. Steve Nelson: If we decide to automatically adopt new RUSP conditions, does that mean we also follow when they decline disorders?
 - 1. Sue Berry: No, if they add something we should add something, but if they decline, as a state we should still have the ability to consider each disease.

- ii. Bob Jacobson: In regards to automatic adoption of the RUSP, does this apply only after the secretary signs off, or when ACHDNC recommends adding condition to the RUSP?
 - 1. Sue Berry: This was discussed. We decided to automatically add a condition once the HHS secretary signs off on the condition, if the secretary turns down the recommended condition, we should still discuss adding it to the MN panel.
- c. Jan (Committee Chair): I propose we discuss in order and then vote after all conditions have been discussed.
 - i. Fabry disease – Work group recommends that we do not add to the panel
 - 1. Discussion
 - 2. Motion was made to have a single vote based on recommendation of the work group.
 - 3. Motion was seconded
 - 4. Motion is amended to have a single vote to follow the recommendations of the work group for inclusion/exclusion of these 7 conditions. To review:
 - a. Fabry: not at this time
 - b. Gaucher: not at this time
 - c. Hurler (MPS-1): pending approval of HHS Secretary
 - d. Krabbe: not at this time
 - e. Niemann-Pick A/B: not at this time
 - f. Pompe: yes, added to RUSP
 - g. X-ALD: pending approval of HHS Secretary
 - ii. Jan (Committee Chair) prompts for more discussion
 - 1. Martha Overby: Brother is a pediatric neurologist in NY, asked his advice based on NY's experience, especially in regards to Krabbe. Brother didn't have expertise but consulted with colleague with experience in treatment and testing. Colleague recommended adding Krabbe to panel. If committee decided to align with work group recommendations, when would we address the other conditions again? Perhaps we should address revisiting this issue later.
 - 2. Dietrich Matern: Federally, not sure when it will be discussed again after reviewing the ACHDNC Evidence Review Group (ERG) results. I expect that the Hunter's Hope Krabbe Newborn Screening Task Force will discuss this in two weeks and fill in the gaps that were identified by the ERG. I believe it will be revisited soon. Anyone can suggest that the ACHDNC committee look into it again.
 - 3. Jan (Committee Chair) invites families to continue to be advocates to help move these conditions forward in the future.
 - 4. Mark McCann: What was a summary of the discussion for and against the work group decisions
 - a. Sue Berry: Krabbe has multiple forms of onset, later onset disorders deserve attention, but have to balance with newborn screening period and how that affects the life of the family. Infantile Krabbe: intervention and effectiveness of treatment. Strategy of how to minimize false positives and the invasiveness

that comes with it. We'd have to have some clear discussions on how to revise NY's screening algorithm.

- b. Gerald Raymond (guest speaker and member of work group): Issues with the diagnostics, need further refinement. High false positive rate which means families run through invasive diagnostic algorithm that they may not need. Also requires a lot of man power for the program and the clinics/hospitals. With the number of individuals who have infantile Krabbe, how many are we likely to see, can we stay competitive and competent with diagnosis and follow-up if the condition is this rare? Could we be missing children as well, even if we screen? How would we do this?
 - c. Chet Whitley (member of work group): I see almost all the LSD in MN, and represent the minority opinion. We need early intervention and treatment. We are already screening for things that don't require treatment, are we going to only screen for disorders with narrow guidelines or are we going to treat patients because in the long run people will benefit. We should screen for Fabry, Krabbe, and Gaucher.
5. Jan (Committee Chair): We have time for comments from the public.
- a. Multiple families spoke about their experiences.
 - b. Annamarie Saarinen (Newborn Foundation): Need clarity for methodology for lack of symptoms in newborn period for assessment tool and criteria for deciding on recommendations. We don't need the legislature. We have the authority to add our suggestions to NBS panel, not sure about waiting for HHS Secretary to sign off on these, it's been a very thorough process in MN and through ACHDNC. Concerned about waiting for HHS Secretary to sign off, it's tenuous and can take a long time. If we are in consensus, we shouldn't have to wait for the HHS secretary to sign off. CCHD had issues in the past because of this. Even waiting 3-4 months, that's a baby. It takes time to implement, why wait even longer? Thank you to the families who have shared their stories and supported this effort.
 - c. Sue Berry: Symptoms aren't detectible in the newborn period. However, onset of symptoms in the ones we questioned are much further out than the newborn period. Krabbe was the most difficult one to decide on regarding utilization and outcomes. Work group wanted to create a clear path forward for future nominations and create a guideline for decision to align.
 - d. Dietrich Matern: There is a practical part to this. If HHS Secretary signs off in December, then we would put it into effect, we wouldn't have to wait for April's meeting to approve these conditions. Screening for X-ALD and MPS-1 uses tandem mass spectrometry just like is used for Pompe. For parents who want testing for Krabbe, Hunter's Hope provides a link to labs that have testing.

6. Steve Johnson: Vote on resolution that is before us, to adopt the recommendation of the subcommittee. If that resolution fails, then we can go into specific action items based on disorder panel list.
 - a. Jan (Committee Chair) Motion has already been seconded, all in favor?
 - i. In Favor: 15
 1. This vote included an absentee ballot that aligned with the work group's recommendation
 - ii. Opposed: 3
 - iii. Motion carries
7. Jan (Committee Chair) Second Motion: Committee approves conditions that are added to the RUSP. If a condition goes on the RUSP, it is automatically recommended to the MN NBS panel and MDH commissioner. We can go beyond the RUSP, but our recommendations would automatically include all RUSP conditions.
 - a. Steve Johnson: Rather than "automatically" we presumptively act that we add to the panel, that we still have action in this process.
 - b. Jan: Yes, I agree.
 - c. Vote:
 - i. In Favor: 17
 - ii. Opposed: 0
 - iii. Motion carries
8. Sue Berry: Third Motion: We recommend the Department of Health ensures 1) comprehensive long term follow-up of children identified as affected by newborn screening to provide a continuing means for determine the optimization of outcomes for diagnosed citizens; 2) increased awareness of the utility of long term follow-up; 3) and improved education to parents and providers about outcomes of newborn screening conditions and their long-term management.
 - a. Candace Lindow-Davies: Second
 - b. Vote:
 - i. In Favor: 17
 - ii. Opposed: 0
 - iii. Motion carries
7. Jan (Committee Chair): Is there any other business?
 - a. Annamarie Saarinen: Something to consider for the next meeting, with the work group committees, can we have a parent as part of the evidence review process? Can we have a formal role for parents in these work groups?
8. Closure
 - a. Jan (Committee Chair): Notified committee that next meeting is in April and thanked everyone for their presence and work.
 - b. Meeting is adjourned.